

## Arthrogryposis multiplex congenita distal type II associated with facial abnormality, renal abnormality, Polydactyly and Hirschprung's disease

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### Summary

A case of arthrogryposis multiplex congenita distal type II associated with facial abnormality, renal abnormality, postaxial polydactyly and Hirschprung's disease is described. It appears to be a new form of an autosomal recessive disorder.

### Introduction

Arthrogryposis is a syndrome involving multiple persistent joint contractures in the intact skeleton at birth. Severity of the condition is variable. It is classified into two types, type I with only distal limb involvement, and type II with other defects. Numerous aetiological causes have been implicated, including genetic. In the genetic variety autosomal recessive, autosomal dominant and X-linked recessive forms of transmission have been reported. The classical form (type I), called amyoplasia (7), is always sporadic.

### Case report

A 22-day old male neonate was admitted to the

General Hospital Peradeniya, Sri Lanka, with abdominal distension and vomiting. He was a product of a consanguineous marriage. The mother was 28 years old at the time of conception. She had an uncomplicated pregnancy. The child was born at full term as a normal vaginal delivery in an estate hospital. A week later the mother noticed gradual distension of the neonate's abdomen. Subsequently, the child started vomiting several times a day, and was constipated for several days at a time.

On examination, the child was not active and the weight was 2300 g, with a crown to heel length of 50 cm. Thorough examination of the child revealed many abnormalities. A thick, rough, hairy and heavily pigmented skin, a prominent wide-open meiotic suture, low hairline, low-set, wide and posteriorly pointing auricles, hypertelorism, micrognathia and a webbed neck (Figure 1); abdomen distended with gas, girth 36 cm, visible veins on the abdominal wall and a high placed anus; in limbs, long proximal and middle phalanges, flexed deformity in both wrists, flexion deformity of both ankles, congenital dislocation of hips, postaxial Polydactyly on the right and congenital talipes equinovarus on the left (Figure 2).

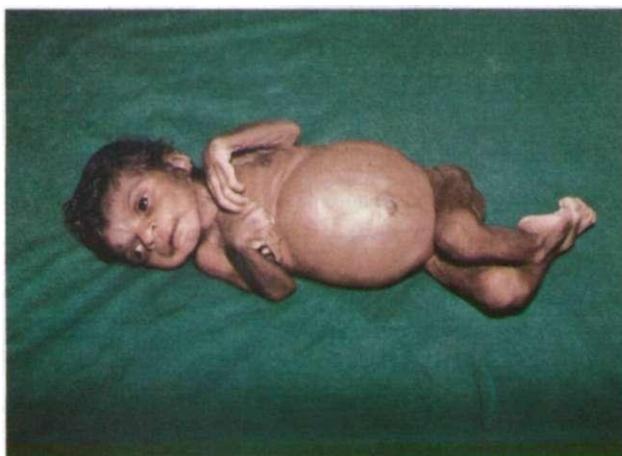


Figure 1. Distended abdomen and limb and facial abnormalities.



Figure 2. Abnormalities of the feet and toes.

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Blood urea was 18.6 mmol /l. Plain radiograph of the abdomen revealed distended bowel loops with gas. Plain radiograph of the pelvic region showed congenital dislocation of hips with underlying slanted acetabular roof, short left femur due to congenital dislocation, absent lower femoral epiphysis, absent rectal gas and distended large bowel loops. Ultrasonography of the abdomen revealed highly echogenic kidneys and gaseous distention of bowel. All other intra-abdominal organs were ultrasonically normal. Transfontanelle ultrasound revealed normal brain parenchyma with a normal ventricular system.

The diagnosis of Hirschsprung's disease was based on the continuing constipation, and severe abdominal distension with large bowel loops not coming down to the rectum seen on the plain abdominal xray. Study of the pedigree confirmed the consanguinity. No history of similar illness was found for the three previous generations investigated.

A geneticist was of the view that the neonate did not have characteristic features of any syndrome due to chromosomal abnormality, and that it could be a variant form of arthrogryposis. A decision was taken not to treat the child, and that it would be unethical to do any invasive investigations.

The infant succumbed at the age of 64 days in the estate dispensary. Unfortunately, the body was not available for an autopsy. After the death of this child the mother conceived again. Following an uneventful pregnancy, she delivered a 2.5 kg girl with no abnormalities. At the time of writing this girl in one year old and has no demonstrable physical abnormalities. Development milestones of this child are normal.

## Discussion

The only previous report in Sri Lanka about arthrogryposis is in 1957 (1). World-wide there are a significant number of case reports of arthrogryposis but none describes the features described in this report.

The picture seen in this neonate exemplifies most of the previously reported findings, but spine or thoracic abnormalities (2) as well as cleft palate (3) and congenital heart disease (4) were not seen. Micrognathia and webbed neck described in distal type II arthrogryposis (3) were seen in this child. Hypertelorism, micrognathia, posteriorly angulated ears are other findings which are similar to previous reports (5). High serum urea level and ultrasonic finding of highly echogenic kidneys are evidence to suggest renal dysfunction in this child (6). The affected infants are usually not mentally retarded (7). The physical findings as well as investigations suggest no abnormalities in the central nervous system.

Hirschsprung's disease has been previously described in two brothers in association with congenital heart mal-

formation, broad big toes and ulnar Polydactyly (8). There is also a recorded case of a brother and sister born to first cousin parents with a syndrome of Hirschsprung's disease, Polydactyly, unilateral renal agenesis, hypertelorism and congenital deafness (9). This child had clinically satisfactory pulmonary function, and did not have evidence of pulmonary hypoplasia (10,11,12).

The neonate that we report appeared to be a having a new form of an autosomal recessive syndrome, with the association of Polydactyly, Hirschsprung's disease and arthrogryposis multiplex congenita distal type II.

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